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Biochemical Disorders and Clinical Applications

Biochemistry

Diabetes mellitus, dyslipidemia, gout, phenylketonuria, glycogen storage disorders, vitamin deficiency disorders and other common metabolic disorders.

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CHAPTER 20

Biochemical Disorders and Clinical Applications

Diabetes, dyslipidaemia, gout, inherited metabolic disorders and deficiencies

1. Diabetes mellitus

Diabetes is a group of disorders characterized by hyperglycaemia due to impaired insulin secretion, action or both. Type 1 usually results from autoimmune beta-cell destruction; type 2 involves insulin resistance and progressive beta-cell dysfunction. Gestational diabetes is diagnosed during pregnancy using pregnancy-specific criteria. Other forms include monogenic, pancreatic and drug-associated diabetes. See Chapter 18 for diagnostic thresholds.

Insulin deficiency or resistance increases hepatic glucose output and reduces effective glucose utilization, causing hyperglycaemia. When renal reabsorptive capacity is exceeded, glycosuria and osmotic diuresis can cause polyuria, thirst and dehydration. Chronic hyperglycaemia, oxidative stress and vascular injury contribute to retinopathy, nephropathy, neuropathy and cardiovascular disease.

Acute complication	Biochemical basis and alert features
Hypoglycaemia	Blood glucose <70 mg/dL is an alert threshold in diabetes care; sweating, tremor, confusion or seizures may occur
Diabetic ketoacidosis (DKA)	Insulin deficiency leads to lipolysis, ketones and metabolic acidosis; vomiting, abdominal pain and deep breathing may occur. Glucose may not be markedly elevated in some cases.
Hyperosmolar hyperglycaemic state (HHS)	Severe hyperglycaemia, hyperosmolality and dehydration; altered mental status may occur

These are clinical emergencies when severe or suspected. Assess, check glucose and escalate according to protocol. Never give oral food or fluid to a patient unable to swallow safely. Lifelong management includes individualized nutrition, medication, monitoring, education and prevention of complications.

2. Dyslipidaemia

Abnormal concentrations of lipoproteins or lipids. Elevated LDL-C, triglyceride-rich remnants and apoB-containing particles contribute to atherosclerosis; severe hypertriglyceridaemia can cause pancreatitis. Familial hypercholesterolaemia often involves impaired LDL receptor pathways. Treatment is risk-based and may include lipid-lowering medication with dietary and lifestyle support. Monitor lipid profile, adherence and clinically important adverse effects.

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3. Gout and purine metabolism

Purines are degraded through hypoxanthine and xanthine to uric acid by xanthine oxidoreductase. Hyperuricaemia may arise from reduced excretion or increased production. Monosodium urate crystal deposition in joints causes gout; stones and tophi may occur. Serum urate can be normal during an acute flare and elevated urate alone is not diagnostic. Allopurinol inhibits xanthine oxidase. Acute red, swollen joints require evaluation, including exclusion of infection where indicated.

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4. Inherited metabolic disorders

Disorder	Biochemical defect	Clinical significance
Phenylketonuria (PKU)	Usually phenylalanine hydroxylase deficiency; phenylalanine accumulates	Untreated elevation can impair neurodevelopment; newborn screening and specialist diet/treatment
Classic galactosaemia	GALT deficiency; galactose-1-phosphate accumulates	Newborn feeding-related illness, jaundice, liver dysfunction and sepsis risk; urgent specialist management
Maple syrup urine disease	Branched-chain alpha-ketoacid dehydrogenase deficiency	Accumulation of leucine and related metabolites; encephalopathy, metabolic crises
Hereditary fructose intolerance	Aldolase B deficiency	Hypoglycaemia and hepatic dysfunction after fructose exposure
G6PD deficiency	Reduced NADPH production in red cells	Oxidant-triggered haemolysis; avoid verified triggers and promptly assess symptoms

5. Glycogen storage disorders

Type	Defect / important features
I (Von Gierke)	Glucose-6-phosphatase system; fasting hypoglycaemia, hepatomegaly, lactate elevation
II (Pompe)	Lysosomal acid alpha-glucosidase; muscle weakness, possible infantile cardiomyopathy
III (Cori/Forbes)	Debranching enzyme; liver and muscle manifestations
V (McArdle)	Muscle glycogen phosphorylase; exercise intolerance and possible rhabdomyolysis

6. Other nutritional and metabolic conditions

Condition	Biochemical basis / signs
Iron deficiency	Insufficient available iron for haem synthesis; microcytic anaemia commonly

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B12 / folate deficiency	Impaired DNA synthesis; megaloblastic changes; B12 can cause neurological symptoms
Vitamin D deficiency	Impaired calcium/phosphate mineralization; rickets/osteomalacia
Scurvy	Vitamin C deficiency impairs collagen function; bleeding and poor healing
Wilson disease	ATP7B-related copper handling defect; hepatic and neurological manifestations
Haemochromatosis	Excess iron accumulation; organ damage over time
Lactic acidosis	Lactate accumulation due to increased production or reduced clearance; evaluate underlying cause

7. Biochemical changes in diabetes: comparison

Feature	Type 1 diabetes	Type 2 diabetes
Main mechanism	Usually autoimmune beta-cell destruction	Insulin resistance and progressive beta-cell dysfunction
Insulin availability	Marked deficiency	Variable; may decline over time
Ketosis	Greater susceptibility when insulin is deficient	Can occur, particularly with severe illness or relative deficiency
Clinical care	Requires insulin replacement	Individualized lifestyle and medication plan; insulin may be needed

Neither age nor body size alone establishes diabetes type. Laboratory and clinical assessment guide classification.

8. Pathways affected in inherited metabolic disorders

Phenylketonuria

Phenylalanine hydroxylase normally converts phenylalanine to tyrosine using tetrahydrobiopterin (BH4). Deficient activity causes phenylalanine accumulation, which can harm neurodevelopment. Newborn screening and early specialist management reduce risk. Dietary phenylalanine restriction requires professional supervision because phenylalanine is also an essential amino acid.

Glycogen storage disease

Defects in glycogen synthesis, degradation or lysosomal handling cause distinct clinical patterns. Liver-predominant disorders may cause fasting hypoglycaemia; muscle-predominant disorders may cause exercise intolerance. Pompe disease is a lysosomal storage disorder involving acid alpha-glucosidase deficiency. Treatment is disease-specific.

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Gout

Uric acid is produced from purine breakdown and is eliminated principally through renal and intestinal routes. Crystal deposition causes inflammation. Hyperuricaemia alone is insufficient to diagnose gout; acute monoarthritis also requires consideration of septic arthritis. Long-term treatment decisions are clinical, not determined by a single urate reading.

9. Nursing responsibilities

Recognize metabolic warning signs (altered consciousness, dehydration, seizures, respiratory changes, vomiting). Follow authorized emergency protocols, collect specimens safely and monitor trends. Support specialist dietary requirements for inherited disorders, respect genetic confidentiality and teach patients when and how to seek urgent care. Ensure families understand follow-up and avoid unprescribed supplements or dietary restrictions.

10. Additional biochemical disorders

Condition	Underlying disturbance / relevance
Lactose intolerance	Low lactase activity; undigested lactose causes osmotic symptoms and bacterial fermentation.
Familial hypercholesterolaemia	Inherited disruption of LDL clearance, often involving LDLR; early atherosclerotic risk.
Urea cycle disorder	Defects in nitrogen disposal may cause hyperammonaemia; acute neurologic changes require urgent specialist care.
Medium-chain acyl-CoA dehydrogenase deficiency	Impaired fatty acid oxidation; fasting or illness may trigger hypoketotic hypoglycaemia.

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Condition	Underlying disturbance / relevance
Lesch-Nyhan syndrome	HGPRT deficiency disrupts purine salvage, causing excess uric acid and neurological manifestations.

11. Clinical case review

Case 1: A patient with diabetes develops vomiting, deep rapid breathing and dehydration. Hyperglycaemia and metabolic acidosis with raised ketones support suspected DKA. The immediate nursing priority is clinical assessment, urgent escalation and monitoring according to the emergency protocol.

Case 2: A patient presents with pallor, fatigue and low haemoglobin with microcytosis. Iron deficiency is one possibility, but thalassaemia and inflammation also require consideration. Review iron studies and clinical history rather than assuming all microcytic anaemia is due to iron deficiency.

Case 3: A newborn becomes lethargic with vomiting and feeding difficulty. Inherited metabolic disease is one possible cause among serious alternatives such as sepsis. Immediate clinical assessment is essential; newborn screening results are not a substitute for urgent evaluation.

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12. Important Examination Questions

Long Answer Questions

1. 1. Discuss biochemical changes in diabetes and common metabolic disorders.
2. 2. Explain the biochemical basis of diabetes, DKA and HHS.
3. 3. Describe phenylketonuria, gout and selected inherited metabolic disorders.

Short Answer Questions

1. 1. Write a short note on DKA.
2. 2. Write a short note on gout.
3. 3. Write a short note on PKU.
4. 4. Write a short note on glycogen storage disease.
5. 5. Write a short note on dyslipidaemia.

Very Short Answer Questions

1. 1. What enzyme is deficient in classic galactosaemia? - Galactose-1-phosphate uridylyltransferase (GALT).
2. 2. Which glycogen storage disorder involves muscle glycogen phosphorylase deficiency? - McArdle disease (type V).
3. 3. What biochemical defect causes G6PD-related vulnerability? - Impaired NADPH generation and reduced antioxidant defence in red cells.
4. 4. What are the three classic symptoms of hyperglycaemia? - Polyuria, polydipsia and unexplained weight loss are classic features; polyphagia may also occur.

Additional objective question

Which enzyme is commonly deficient in PKU? - Phenylalanine hydroxylase.

13. Quick Revision

- Insulin deficiency or resistance produces abnormal glucose regulation.
- DKA involves excess ketones and metabolic acidosis; HHS involves marked hyperosmolality and dehydration.
- PKU commonly results from phenylalanine hydroxylase deficiency.
- Gout is associated with monosodium urate crystal deposition.

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Conclusion

Biochemical disorders illustrate how disrupted enzymes, transport pathways and hormonal regulation produce recognizable clinical patterns that guide safe assessment and referral.

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Practice questions

Choose one best answer. The answer key and explanations follow the questions.

1. Which crystal deposition causes gout?

A. Calcium oxalate

B. Cholesterol

C. Calcium phosphate

D. Monosodium urate

2. Classic galactosaemia usually results from deficiency of which enzyme?

A. Aldolase B

B. GALT

C. Phenylalanine hydroxylase

D. Glucose-6-phosphatase

3. Which biochemical disturbance characterizes diabetic ketoacidosis?

A. Ketone production with metabolic acidosis

B. Marked hyperosmolality without significant ketone acidosis

C. Low glucose with adrenergic symptoms

D. High urate with joint crystal deposition

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Answer key and explanations

1. D - Monosodium urate

Gout results from monosodium urate crystal deposition in joints.

2. B - GALT

Classic galactosaemia is caused by GALT deficiency with galactose-1-phosphate accumulation.

3. A - Ketone production with metabolic acidosis

Insulin deficiency promotes lipolysis, ketones and metabolic acidosis.

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